Book Reviews

Am. J. Hum. Genet. 72:1353, 2003

Molecular Cytogenetics: Protocols and Applications. Edited by Yao-Shan Fan. Totowa, New Jersey: Humana Press, 2002. Pp. 411. $135.

In 1986, Pinkel and Gray first utilized FISH techniques in cytogenetic diagnoses. FISH technology, a combination of molecular and conventional cytogenetic techniques, has brought modern cytogenetics to a new era with significantly higher resolutions and a much wider testing spectrum. Since then, numerous new FISH-based technologies have been emerging, from metaphase FISH to interphase FISH and then fiber FISH, from single-color FISH to spectral karyotyping (SKY) and multicolor FISH (M-FISH), from regular FISH to reverse FISH (also called "microFISH"), from comparative genomic hybridization (CGH) to microarray CGH, and so on. Each technique is associated with a specific testing spectrum and limitations. It is neither easy for clinicians to choose the right tests for their patients—i.e., those that will bring the benefits afforded by the new technologies—nor straightforward for clinical cytogeneticists to decide which new technologies to implement in their laboratories with limited laboratory budgets and low reimbursement rates.

Although many books and articles about FISH technologies and their clinical applications have been published, a book that contains step-by-step and easy-to-follow protocols for most of the new FISH technologies was not available until the recent publication of *Molecular Cytogenetics: Protocols and Applications,* edited by Yao-Shan Fan. Dr. Fan has done a marvelous job of putting together a collection of laboratory protocols and review articles of different FISH technologies. The book is divided into four parts: (1) "Basic Concepts and Techniques," including FISH probe sources, fluorophores, probe labeling, and basic FISH procedures; (2) "Evolving Techniques and Applications," including chromosome microdissection (microFISH), primed in situ labeling (PRINS), SKY, M-FISH, CGH, color banding, fiber FISH, telomere FISH, and microarray CGH; (3) "Special Applications in Chromosomal Disorders," including delineating marker chromosomes, prenatal diagnosis of common aneuploidies, prenatal diagnosis using fetal cells in maternal blood, preimplantation test, microdeletion syndromes, uniparental disomy, and reproductive pathology; and (4) "Special Application in Oncology," including FISH in chronic myeloid leukemia (CML), FISH detecting Her-2/neu amplification in breast cancer, CGH in cancer study, etc.

The book focuses mainly on laboratory diagnosis. Most of the chapters were written in a format similar to a laboratory procedure manual, with step-by-step instructions. Some chapters are especially well written and deserve special mention: chapters 7, 8, 9, 12, and 18, describing SKY, M-FISH, CGH, multitelomere FISH, and preimplantation tests, respectively, are of exceptional value for clinical cytogenetic laboratory directors and technologists. The authors of the different chapters not only gave detailed, easy-to-follow protocols, but also provided their first-hand experiences of troubleshooting. Chapter 19 concisely summarizes common microdeletion and microduplication syndromes, a good reference for neonatologists and dysmorphologists. Although the book targets diagnosis, some chapters can be useful for researchers in the fields of human development and cancer biology, such as microdissection and microarray CGH.

The most apparent drawback of this book is that it does not emphasize enough FISH applications in cancer genetics. According to the recently published *The World Health Organization Classification of Neoplasms,* one of the most important biological features used for the classification of cancer is the cytogenetic alteration of tumor cells. These alterations are directly associated with patient responses to therapies and are therefore of not only diagnostic but also prognostic significance. In many cytogenetic laboratories, FISH has become a major diagnostic method for hematopoietic malignancies and solid tumors. In this regard, *Introduction to Fluorescence In Situ Hybridization, Principles and Clinical Applications,* by Andreeff and Pinkel, would be an excellent reference for cytogeneticists as well as hematologists and oncologists, although it does not include step-by-step protocols, as does Dr. Fan's book.

We all understand that there is no book that includes everything about molecular cytogenetics. Compared with other related books, *Molecular Cytogenetics: Protocols and Applications* is packed with information on cutting-edge FISH technologies and their applications, limitations, and pitfalls, along with detailed laboratory protocols. I would certainly recommend that this book be included in the libraries of cytogeneticists and cytogenetic laboratories.

MARILYN M. LI

Hayward Genetics Center
Tulane University School of Medicine
New Orleans

0002-9297/2003/7205-0034$15.00

Am. J. Hum. Genet. 72:1354, 2003

Down Syndrome: Visions for the 21st Century. Edited by William I. Cohen, Lynn Nadel, and Myra E. Madnick. New York: John Wiley and Sons, 2002. Pp. 473. $27.50.

This is a multiauthored book edited by three international experts on Down syndrome. The authors were presenters at a National Down Syndrome Society conference held July 27–29, 2000, in Washington, D.C.

The book is divided into 10 parts and is directed to the interests of parents as well as professionals. This is indeed a very challenging endeavor. The first four parts address self-determination, self-advocacy, advocacy, and the role of the family. The largest part of the book is on health and clinical care and covers such topics as advances in pediatric health and health care guidelines in Down syndrome, physical therapy, behavioral concerns, and life issues for adolescents and adults. The part of the book that is focused on research includes a detailed description of the sequence of chromosome 21, the origin and etiology of trisomy 21, the genetic origins of cognition and heart disease, and nonconventional therapies for Down syndrome. The last parts of the book, parts seven, eight, and nine, deal with psychosocial issues, education/inclusion, and communication, math and language skills. The final part of the book addresses visions for the individual with Down syndrome and the reality and challenges for the future.

In general, the book does a very nice job of balancing important and technical information for parents, as well as providing the latest updates for professionals on health, clinical, and research issues on Down syndrome. It brings into focus issues on behavior and postsecondary education and updates an ongoing dialogue on speech/language and education of individuals with Down syndrome. It provides an excellent review of nonconventional therapies for Down syndrome.

In summary, this book is exceedingly valuable for a wide spectrum of individuals: parents, professionals, caregivers, and researchers on Down syndrome. It does a particularly good job in updating new knowledge on key topics pertinent to individuals with Down syndrome.

DON C. VAN DYKE
Divisions of Developmental Disabilities and Medical Genetics
University of Iowa Health Care
Iowa City

0002-9297/2003/7205-0035$15.00